```
=> d his

     (FILE 'HOME' ENTERED AT 18:20:59 ON 09 FEB 2003)

     FILE 'HCAPLUS' ENTERED AT 18:21:11 ON 09 FEB 2003
                E SITOSTEROLEMIA SUSCEPTIBILITY GENE/CT
L1              1 S SITOSTEROLEMIA SUSCEPTIBILITY GENE
                E ABC TRANSPORTER/CT
                E E4+ALL
                E E2
                E E3+ALL
L2            417 S E3
L3             75 S SITOSTEROLEM?
L4              1 S L2 (L) L3
```

```
=> e e3+all
E1      609132   BT2  Proteins/CT
E2       19180     BT1  Transport proteins/CT
E3                   --> Transport proteins (L) ABC (ATP-binding cassette)
                         transporters/CT
E4                     OLD  Proteins, specific or class (L) ABC (ATP-binding
                            cassette-contg.)/CT
E5                     OLD  Transport proteins (L) ABC (ATP-binding
                            cassette-contg.)/CT
E6                     UF   ABC proteins/CT
E7                     UF   ABC transporters/CT
E8                     UF   ATP-binding cassette transporters/CT
E9                     UF   ATP-binding cassette-contg. proteins/CT
```

=> d ibib ab 1

L4 ANSWER 1 OF 1 HCAPLUS COPYRIGHT 2003 ACS
ACCESSION NUMBER: 2002:135503 HCAPLUS
DOCUMENT NUMBER: 136:399968
TITLE: Novel donor splice site mutation of ABCG5 gene in sitosterolemia
AUTHOR(S): Lam, Ching-Wan; Cheng, Anna Wai-Fun; Tong, Sui-Fan; Chan, Yan-Wo
CORPORATE SOURCE: Department of Medicine, Brigham and Women's Hospital, Boston, MA, 02115, USA
SOURCE: Molecular Genetics and Metabolism (2002), 75(2), 178-180
CODEN: MGMEFF; ISSN: 1096-7192
PUBLISHER: Academic Press
DOCUMENT TYPE: Journal
LANGUAGE: English
AB In a patient with sitosterolemia, we found two different mutations of the ATP-binding cassette, subfamily G, member 5 (ABCG5) gene. The first is a missense mutation that changes the amino acid residue at position 419 from arginine to histidine, i.e., R419H. The second is a novel splicing mutation affecting the invariant guanine at the first base of the donor splice site of intron 12, i.e., IVS12+1G A. The father of the patient is heterozygous for the missense mutation, and the mother is heterozygous for the splicing mutation. No mutations were found in the sister of the patient. Up until now, the missense mutation has only been found in Japanese patients with sitosterolemia. We believe that R419H in our Chinese patient may have the same origin as the mutation in the Japanese patients with sitosterolemia. (c) 2002 Academic Press.
REFERENCE COUNT: 4 THERE ARE 4 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT